counselling sessions with her family revealed that there were at least four of her relatives suffering from the same symptoms. We draw a pedigree displaying three generations and consanguinity of the family and carried out a WES analysis to selected. Results revealed that, three severely affected family members had 1-bp insertion in the *WNK1* gene and homozygous for the allele. This *WNK1* gene was one of the candidate genes for HSAN type 2.

https://doi.org/10.1016/j.jbiotec.2018.06.062

Gene variants of Congenital Adrenal Hyperplasia in Anatolian population

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Congenital adrenal hyperplasia (CAH) refers to a group of several autosomal recessive diseases resulting from alteration of genes for enzymes mediating the biochemical steps of production of the adrenal gland hormones. Classic form of CAH represents the severe form while the non-classic form representing the milder and more common form of CAH. 21-hydroxylase, 3-beta-hydroxysteroid dehydrogenase, 11-beta-hydroxylase, 17-alpha-hydroxylase deficiencies are known to be associated with CAH. In our study, we investigated *CYP21A2*, *CYP11B1*, *HSD3B2* genes which are associated with 21-hydroxylase, 11-beta-hydroxylase and 3-beta-hydroxysteroid dehydrogenase enzyme deficiencies, respectively, in 365 individuals. Sanger sequencing method was used for investigation of genes of interest. 239 (65%) patients with non-classic CAH, 51 (14%) patients with classic simple virilizing CAH, 46 (13%) patients with classic salt wasting CAH and 29 (8%) individuals with family history of CAH were evaluated. 1 or more than 1 variants were detected in 269 of 365 (74%) individuals including 161 of 239 (67%) patients with non-classic CAH, 44 of 51 (86%) patients with classic simple virilizing CAH, 43 of 46 (93%) patients with classic salt wasting CAH and 21 of 29 (72%) individuals with family history of CAH. 32 variants of *CYP21A2* including 10 novel variants, 9 variants of *CYP11B1* including 3 novel variants and 6 variants of *HSD3B2* including 4 novel variants were detected. Our results indicate that in Anatolia, discovery of novel mutations is still common due to high rates of consanguineous marriages which increase the frequency of CAH.

https://doi.org/10.1016/j.jbiotec.2018.06.063

Do UCP2, IL-17, mi196a2, and *NR3C1* gene variants contribute to the risk of microtia? A preliminary study in Turkish population

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Microtia is a congenital malformation of variable severity of the external and middle ear. Although many genetic and environmental factors are investigated the etiopathogenesis of microtia, it is still uncertain. We performed case–control analysis to assess the effect of UCP2 −866 G/A, IL-17 −7488 A/G, miR196a2 T/C, NR3C1 Bcl1 variants on the risk of microtia. This study includes 18 microtia patients and 70 healthy controls. The functional variants of UCP2, IL-17, miR196a2, and *NR3C1* genes were evaluated using PCR-RFLP method. The frequencies of the alleles and genotypes in groups were compared by the χ^2 test A significant difference was found between the control group and the patients as for genotype and allele frequencies of NR3C1 Bcl1 variant. No significant difference was found in the distribution of genotypes and alleles frequencies between patients and healthy controls for UCP2, IL-17, and miR196a2 variants ($p > 0.05$). To our knowledge, this is the first study to evaluate relationship between these gene variants and microtia risk in a Turkish cohort. Our results suggest that the *NR3C1* Bcl1 variant might reflect the risk of microtia in a Turkish population. Further studies in larger populations are required to achieve a definitive conclusion.

https://doi.org/10.1016/j.jbiotec.2018.06.064